Radiology Quiz

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Clinical History

A 14-year-old Saudi boy started to have generalized body weakness and abnormal movement in both legs. For the last 6 weeks, he became bedbound. Recently the patient presented with severe dysarthria.

Questions

1. What are the radiographic findings?
2. What is the diagnosis?
Figure 1 - Axial T2 WI (TR/TE 4000/115) at the level of middle cerebellar pedunele, shows high signal intensity at the dentate nuclei and to a lesser degree at the basis pontis.

Figure 2 - Axial T2 WI C TR/TE 4000/115 at the level of lateral ventricle, shows high signal intensity at both thalamic and lentiform nuclei bilaterally and symmetrically.

Figure 3 - Axial T2 (TR/TE 4000/115) at the level of mid brain shows high signal intensity affecting the mid brain and substantia nigra.

Figure 4 - Axial T2 WI (TR/TE 4000/115) at the level of periventrical region shows high signal intensity at the subcortical region of the left frontal lobe. Note also the prominence of the CSF space over the cerebral convexities due to a mild degree of cortical atrophy.
Axial T2 WI (TR/TE 4000/115) of the brain (Figure 1) demonstrates a high signal intensity affecting the dentate nuclei bilaterally and to a lesser extent at the basis pontis. Bilateral and symmetrical hyper intensities are seen in both the thalamic nuclei and lenti form, caudate atrophy is also noted (Figure 2).

There is high signal intensity at the mid-brain, substansia nigra and at the left hippocampus with subsequent dilatation of the ipsilateral temporal horn and to a lesser extent at the right hippocampus (Figure 3). Additionally, high signal intensity at the subcortical region of the left frontal lobe with dilatation of the adjacent CSF space due to a mild degree of cortical atrophy (Figure 4).

**Diagnosis**

The differential diagnostic possibilities of bilateral symmetrical basal ganglia changes and white matter abnormalities include:

1. Wilson Disease
2. Osmotic myelinolysis with extrapontine sites
3. Toxic encephalopathy (alcohol)
4. Vasculitis
5. Hypoxic - ischemic insult

The patient was found to have abnormal liver function tests with high copper identified in the serum and urine.

**Final diagnosis**

Wilson’s Disease.

**Discussion.** Wilson’s disease hepato-lentricular degeneration, was first described by Kinner Wilson in 1912 and is a genetically determined autosomal recessive inherited disorder of copper metabolism characterized by abnormal deposition of copper in multiple organs including the liver as primary copper storage organ, brain, cornea, kidney and bones due to ceruloplasmin - the serum transport protein of copper deficiency.

The defective gene is on the long arm of chromosome 12. Although Wilson’s disease can occur at any age, most affected patients become symptomatic between the age of 8 and 16 years. It may be manifested by hepatitis or liver failure which may be acute and massive or slow as cirrhotic liver, however, most patients, whatever their clinical presentation, have some degree of liver disease. The neurological presentation depends on the area involved which occurs relatively later in life in comparison to the hepatic presentation - manifested clinically by tremor, dysarthria, dystonia, rigidity, bradykinesia or by psychological symptoms. The pathophysiology of the neurological abnormalities could be due to direct copper deposition in the brain or indirectly as a sequence of liver dysfunction (hepatic encephalopathy) or both. Other clinical manifestations would include Kayser Fleischer ring, which is due to deposition of copper at descement’s membrane at the cornea that is pathognomic for Wilson’s disease.

Definite diagnosis is based on reduced serum cerloplasmin and copper level, increased 24 hour urinary copper excretion and copper analysis of liver biopsy as well as slit-lamp examination for Kayser-Fleischer ring.
Wilson’s disease is suggested by neuroimaging studies mainly T2WI by brain atrophy (focal/diffuse), high signal intensity at the lentiform nuclei with bilateral symmetrical and concentric lamellar putaminal pattern, thalamic and caudate nuclei, substantia nigra, periqueductal grey matter, pontine tegmentum, cerebellum, cortical and subcortical areas with frontal lobe predilection. The high signal intensity could be due to edema, gliosis, demyelination, necrosis, sponge degeneration and cavitory disintegration. Non-enhanced CT may show bilateral putamin low density or generalized brain atrophy. The MR imaging has proved to be more sensitive than CT, not only for diagnosis but some studies show that there is a correlation between the MRI finding and clinical presentation severity. Other studies show that it is a useful tool for evaluation of the treatment.

The initial treatment approach is life long removal of excessive copper by chelating agent such as pencillamine or zinc acetate, which has a lower toxicity.

References